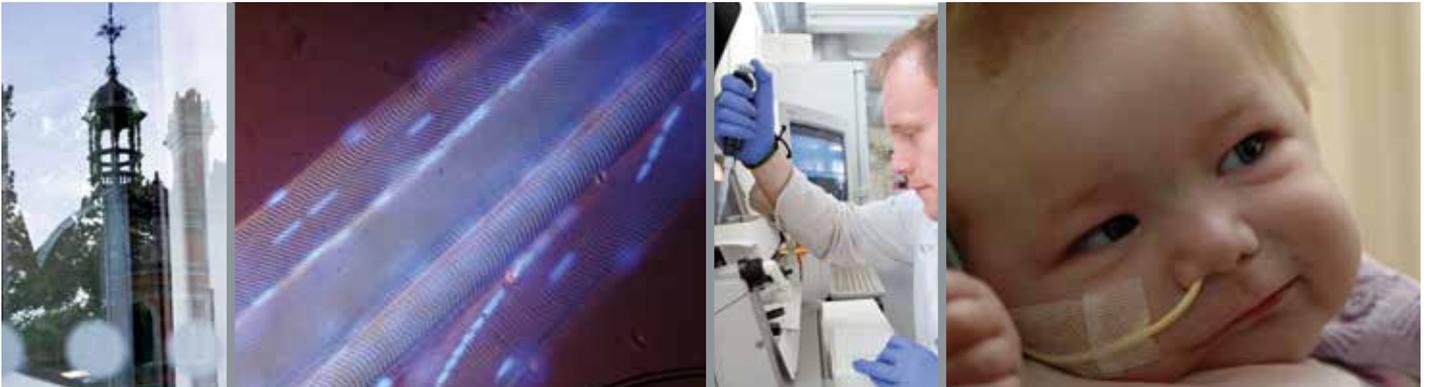




Rare Disease Day

Rare disorders without borders!



Thursday 28 February 2013

**A day of talks, activities and tours
hosted by Royal Holloway**

Programme of events

Rare Disease Day



Rare disorders without borders!

In Europe, a rare disease is defined as one with an incidence of less than 1 in 2,000 people. While each of them is rare, there are more than 7,000 rare diseases, which together affect 24 million (6%) people in Europe and take 20% of all health care costs. Rare diseases are a serious public health concern and an international priority but unfortunately often very little is known about them by health professionals and the public alike.

The aim of Rare Disease Day is to highlight the need for more research and funding to help sufferers and their families and to draw attention to rare diseases and the millions of people who are affected by them.

Rare disease research at Royal Holloway

Royal Holloway is a leading institution in the development of novel therapies for rare diseases, including Spinal muscular atrophy, Spinal injury, Duchenne muscular dystrophy and Severe combined immunodeficiency (referred to as 'child in a bubble' disease). Our scientists are at the forefront in developing innovative techniques in gene therapy.

Visit our Rare Disease Day webpages at www.rhul.ac.uk/rarediseaseday

Organisers

Dr Rafael Yáñez, lead organiser, and Professor George Dickson, co-organiser, School of Biological Sciences, Royal Holloway, University of London.

The organisers wish to thank Royal Holloway's Alumni Fund, Outreach Competitive Fund and Research and Enterprise for sponsoring the event.



Exhibitors and contributors

- Muscular Dystrophy Campaign
- The Jennifer Trust for SMA
- Genetic Alliance UK
- Rare Disease UK
- AKU Society
- DEBRA
- Shine
- Tuberous Sclerosis Association
- Ehlers-Danlos Support UK
- Myasthenia Gravis Association
- Ichthyosis Support Group
- The Society of Biology
- The Association of the British Pharmaceutical Industry
- Genetic Disorders UK
- The British Society for Gene and Cell Therapy
- The SMA Trust
- Ashford and St Peter's Hospitals NHS



"Royal Holloway is at the forefront of research into new therapies for rare diseases and I am delighted that your hard work continues

to be recognised in this way. This Government is committed to providing the best quality of care for people with rare disease. Last year, the four UK health departments launched a consultation on the UK Plan for Rare Diseases, which set out a number of proposals designed to improve co-ordination of care and lead to better outcomes for people with rare disease. This will be the first time that the UK has ever developed a plan on tackling rare disease and I believe these proposals will benefit patients and help the NHS to be more effective in treating these conditions." Rt Hon Philip Hammond

MP, MP for Runnymede & Weybridge and Secretary of State for Defence

Morning session

Picture Gallery, 10am-1pm

- **Genetics through the ages**

With puzzles inspired by Mendel, uncover the recipe of life with Avery, play with the structure of DNA with Franklin all the way through to the human genome project and the future of genetics. This is an opportunity to get hand-on with genetics, the history and the amazing scientists who got us to where we are now.

Hosted by the [Society of Biology](#)

- **Drugs and diseases**

Learn about gene mutations through a Countdown-style spelling game; see if you can match up rare diseases, the genetic variation responsible and the drugs to treat them.

Hosted by the [Association of the British Pharmaceutical Industry](#)

- **DNA origami**

Crack the DNA code and have a go at building your own giant double helix with the help of the British Society for Gene and Cell Therapy and Royal Holloway Biological Science students!

- **Exhibition**

Learn about rare genetic diseases from our representative charities. See our exhibitor list on page 2 for further details.

Afternoon session

Bourne Building and Queen's Building 1-2pm and 2-3pm

The following talks will be live streamed at 1pm (accessed via www.rhul.ac.uk/rarediseaseday)

- An introduction to Rare Disease Day, Dr Rafael Yáñez, School of Biological Sciences
- Living with Neurofibromatosis, Adam Pearson, Patron of Jeans for Genes Day
- Zombie Science, Doctor Austin
- Gene therapy research at Royal Holloway, Professor George Dickson, School of Biological Sciences

Plus!

Quiz our undergraduate students 'speed dating' style (pre-booked slots only)



 @RHULScience

The following activities will run in pre-booked slots.

- **Bioethics of genetic screening**

What are the implications of genetic testing for individuals and their families? Come and find out more about the ethics of rare disease diagnosis and treatment.

Hosted by [Dr Felicity Boardman, University of Warwick](#)

- **Living with genetic disorders**

A video-based discussion activity: how do genetic disorders such as Huntington's and cystic fibrosis impact on sufferers' lives and how are they managed?

Hosted by [Genetic Disorders UK](#)

- **Rare diseases quiz**

Royal Holloway students guide you through some online resources from our genetics teaching modules and other websites. Then test your knowledge with our genetics quiz.

- **Laboratory tours**

Come and take a guided tour around our scientists' labs where we carry out our research into rare diseases.





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